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Chronic granulomatous disease of childhood: an unusual cause of recurrent uncommon infections in a 61-year old individual

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Chronic granulomatous disease (CGD) is a rare congenital immunodeficiency that affects 1:250,000 individuals and is characterized by recurrent bacterial and fungal infections as well as by granuloma formation. We investigated a 61-year old individual who had been suffering from a relapsing skin rash affecting various body regions and manifesting with mildly pruritic and erythematous plaques for the past 20 years. Examination of biopsies obtained from lesional skin revealed a suppurative granulomatous process. Tissue cultures grew *Aspergillus nidulans* and *Aspergillus fumigatus* (confirmed by PCR-based analysis). *Aspergillus nidulans* has often been associated which CGD. Leukocyte function tests and Western blot analysis supported this diagnosis. Direct DNA sequencing led to the identification of a hemizygous missense novel mutation in *CYBB* (c.907C>T) that predicts a p.His303Tyr amino-acid substitution in gp91-*phox*, thus definitely confirming a diagnosis of CGD. We describe the diagnosis of a rare inherited immunodeficiency - chronic granulomatous disease (CGD) - in a 61-year old male, suggesting that mild forms of usually fatal immunodeficiencies should be considered when assessing the occurrence of unusual infectious diseases in apparently healthy individuals.